## **AGENDA**

# **SFARI**

# Spring 2022 Investigator Meeting

New York City Monday - Wednesday, April 11-13

WiFi network: **SimonsGuests**Password: **simonsnyc** 

## Monday, April 11

	Kelsev C. Martin, M.D., Ph.D. (SFARI)
	<b>David Spergel, Ph.D.</b> (Simons Foundation)
9:00 - 9:10 am	Welcome and introduction
8:00 – 9:00 am	Breakfast

#### Session 1 Genetic causes of autism and related neurodevelopmental disorders

Live from Ingrid Daubechies Auditorium, 162 Fifth Avenue

Streaming from Gerald D. Fischbach Auditorium, 160 Fifth Avenue

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9:10 — 9:25 am	Session introduction Alan Packer, Ph.D. (SFARI)
9:25 — 9:50 am	Michael Talkowski, Ph.D. (Massachusetts General Hospital; Broad Institute of MIT and Harvard) Insights into the allelic architecture of autism spectrum disorder from rare coding variation
9:50 - 10:15 am	Yufeng Shen, Ph.D. (Columbia University)
	Integrating de novo and inherited variants in over 42,607 autism cases identifies new risk genes with moderate effect
10:15 – 10:40 am	Kavitha Sarma, Ph.D. (The Wistar Institute)
	Elucidating the consequence of R-loop accumulation in autism spectrum disorders
10:40 - 11:10 am	Coffee break
11:10 – 11:35 am	Elise Robinson, Sc.D. (Harvard T.H. Chan School of Public Health)  Statistical and functional convergence of common and rare variant risk for neuropsychiatric disease at chromosomes 16p and 22q
11:35 – 12:00 pm	<b>Beate St Pourcain, Ph.D.</b> (Max Planck Institute for Psycholinguistics)  Polygenic pleiotropy within ASD and ADHD genetic architectures: Shared risk alleles with discordant polygenic effects
12:00 – 12:30 pm	Panel discussion Moderator: <b>Daniel Geschwind, M.D., Ph.D.</b> (University of California, Los Angeles)
12:30 – 2:00 pm	Lunch and Bridge to Independence fellows networking lunch

#### Session 2 Homeostatic plasticity and sleep in neurodevelopmental disorders

Live from Gerald D. Fischbach Auditorium, 160 Fifth Avenue Streaming from Ingrid Daubechies Auditorium, 162 Fifth Avenue

	2:00 pm	Session introduction
		Brigitta Gundersen, Ph.D. (SFARI)
2:15	– 2:40 pm	Graeme Davis, Ph.D. (University of California, San Francisco)
		Homeostatic plasticity: A biochemical mechanism of brain resilience relevant to the etiology and treatment of neurodevelopmental and neurological disorders
2:40	– 3:05 pm	Graham Diering, Ph.D. (University of North Carolina at Chapel Hill)
		Developmental sleep disruption as a risk factor in autism spectrum disorder
3:05	– 3:35 pm	Coffee break
3:35	– 4:00 pm	Shinjae Chung, Ph.D. (University of Pennsylvania)
		Neural mechanisms underlying sleep disturbances in autism spectrum disorder
4:00	– 4:30 pm	Panel discussion Moderator: <b>Kevin Bender, Ph.D.</b> (University of California, San Francisco)

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4:30 – 4:45 pm	Closing remarks
4:45 – 5:15 pm	Cocktail reception and clinical and cognitive science social
	2nd floor promenade at 160 Fifth Avenue
5:15 – 5:45 pm	Introduction and highlights from 2021 Human Cognitive and Behavioral Science RFA awardees
	Gerald D. Fischbach Auditorium, 160 Fifth Avenue
5:45 – 6:15 pm	Cocktail reception and clinical and cognitive science social
	2nd floor promenade at 160 Fifth Avenue
6:15 – 8:00 pm	Dinner Black Barn

## Tuesday, April 12

8:00 - 9:00 am Breakfast

#### Session 3 Gene expression, cells and behavior

Live from Ingrid Daubechies Auditorium, 162 Fifth Avenue Streaming from Gerald D. Fischbach Auditorium, 160 Fifth Avenue

9:00 – 9:15 am	Session introduction  Julia Sommer, Ph.D. (SFARI)
9:15 — 9:40 am	John Rubenstein, M.D., Ph.D. (University of California, San Francisco) Sequential functions of ASD risk gene Tbr1 in mouse cortical cell fate
	specification and synapse formation: Phenotypic rescue by augmenting WNT signaling
9:40 - 10:05 am	Beatriz Rico, Ph.D. (Kings College London)
	Synapse-specific regulation of protein synthesis for cortical wiring
10:05 – 10:35 am	Coffee break
10:35 – 11:00 am	<b>Renata Batista-Brito, Ph.D.</b> (Albert Einstein College of Medicine)  Mef2c-mediated developmental dysfunction of parvalbumin-expressing interneurons
11:00 – 11:25 am	<b>Hirofumi Morishita, M.D., Ph.D.</b> (Icahn School of Medicine at Mount Sinai) Role of autism risk genes in frontal-thalamic projections underlying social processing in mice
1:25 am — 12:00 pm	Panel discussion Moderator: <b>Mriganka Sur, Ph.D.</b> (Massachusetts Institute of Technology)
12:00 - 1:20 pm	Lunch and genomics of ASD networking lunch

## Session 4 Human research studies

11:

Live from Gerald D. Fischbach Auditorium, 160 Fifth Avenue Streaming from Ingrid Daubechies Auditorium, 162 Fifth Avenue

1:20 — 1:35 pm	Session introduction <b>Paul Wang, M.D.</b> (Clinical Research Associates)
1:35 — 2:00 pm	Amina Abubakar, Ph.D. (The Kenya Medical Research Institute (KEMRI)-Wellcome Trust Research Programme and Aga Khan University)  Challenges and opportunities in carrying out research on neurodevelopmental disorders in Sub-Saharan Africa: Lessons from the Neurodev Study
2:00 – 2:25 pm	<b>Gaspar Taroncher-Oldenburg, Ph.D.</b> (Simons Foundation)  Microbiome and other multi-scale associations along the gut-brain axis in autism

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2:25 – 2:50 pm	Rachel Kelly, Ph.D. (Harvard Medical School) Integrative metabolomic endotyping of autism spectrum disorders
2:50 - 3:10 pm	Coffee break
3:10 – 3:35 pm	John Murray, Ph.D. (Yale University)  Mapping brain-behavior relationships in autism
3:35 – 4:05 pm	Panel discussion Moderator: TBD
4:05 – 4:30 pm	Coffee break
4:30 – 5:30 pm	Keynote panel What's next in ASD research: Perspectives from ASD parent-scientists Live from Gerald D. Fischbach Auditorium, 160 Fifth Avenue Streaming from Ingrid Daubechies Auditorium, 162 Fifth Ave. Panelists: Ted Abel, Ph.D. (University of Iowa) Michael Boland, Ph.D. (Columbia University) Pamela Feliciano, Ph.D. (SPARK) Soo-Kyung Lee, Ph.D. (University at Buffalo) Moderator: Alice Luo Clayton, Ph.D. (SFARI)
5:30 – 5:35 pm	Closing remarks
5:35 – 7:00 pm	Cocktail reception with demo and poster session  2nd floor promenade at 160 Fifth Avenue
7:00 – 8:45 pm	Dinner KYMA

## Wednesday, April 13

8:00 – 9:00 am Breakfast

## **Session 5** Emerging therapeutic opportunities

12:00 - 1:00 pm

Live from Ingrid Daubechies Auditorium, 162 Fifth Avenue

Streaming from Gerald D. Fischbach Auditorium, 160 Fifth Avenue

9:00 — 9:15 am	Session introduction John Spiro, Ph.D. (SFARI)
9:15 – 9:40 am	Arkady Khoutorsky, D.V.M., Ph.D. (McGill University)
	The integrated stress response pathway controls autism-related features a cell-type-specific manner
9:40 – 10:05 am	<b>Allison Bradbury, Ph.D.</b> (Nationwide Children's Hospital; The Ohio State University)
	Optimization and validation of gene therapy using patient specific in vitro and in vivo models of SLC6A1 related autism disorder
10:05 - 10:35 am	Coffee break
10:35 - 11:00 am	Christopher Ahern, Ph.D. (University of Iowa)
	Molecular spell-checking with tRNA to correct stop codon-opathies
11:00 – 11:25 am	Richard Huganir, Ph.D. (Johns Hopkins University)
	Regulation of synaptic plasticity by SynGAP
11:25 – 11:55 am	Panel discussion Moderator: <b>Joseph Gleeson, M.D.</b> (University of California, San Diego)
11:55 am — 12:00 pm	Closing remarks <b>Kelsey C. Martin, M.D., Ph.D.</b> (SFARI)

Lunch and departures